

Ontological Spring
Naumburg, Germany - April 17-20, 2002

What needs to be represented in a biomedical ontology?



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Outline

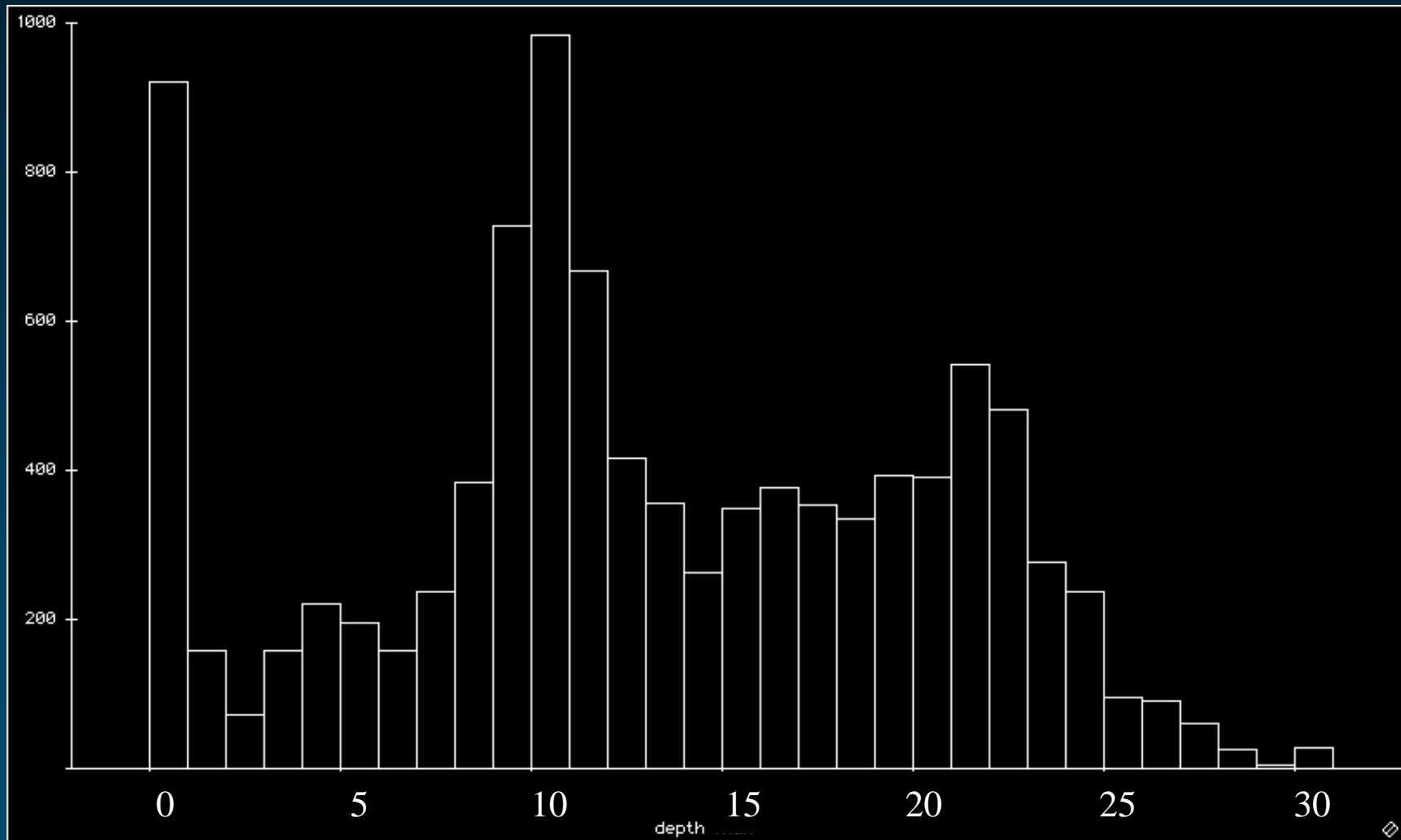
- ◆ Granularity issues
- ◆ Types vs. instances
- ◆ Ontology vs. information model

Granularity issues

Granularity

- ◆ No theoretical limit
- ◆ No ideal granularity
- ◆ Arbitrary limitation
 - Number of digits in code
 - By design (purpose driven)
- ◆ Increased
 - When several perspectives are combined (UMLS / individual vocabularies)
 - When “hierarchy” is loosely defined

Granularity in UMLS

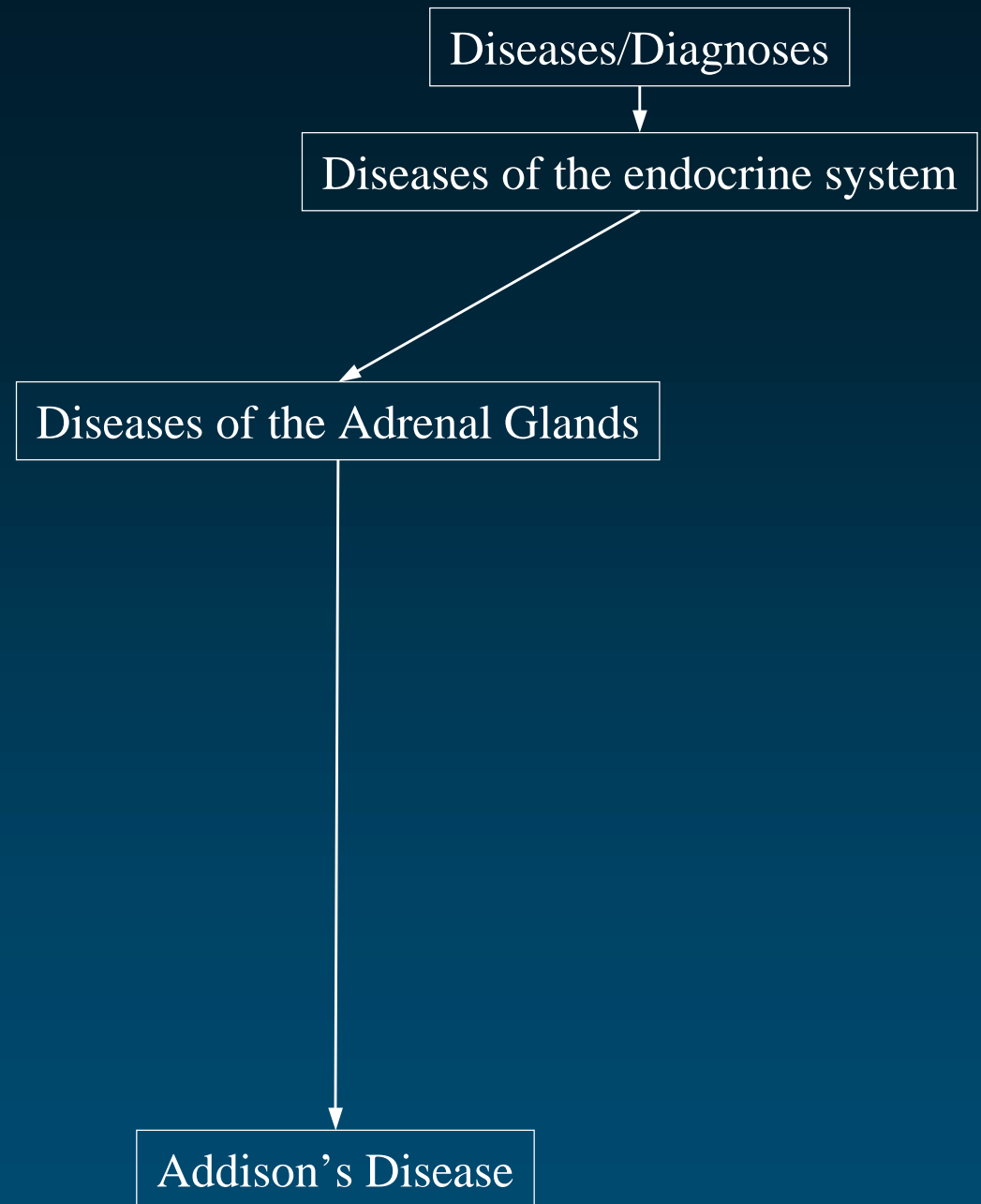


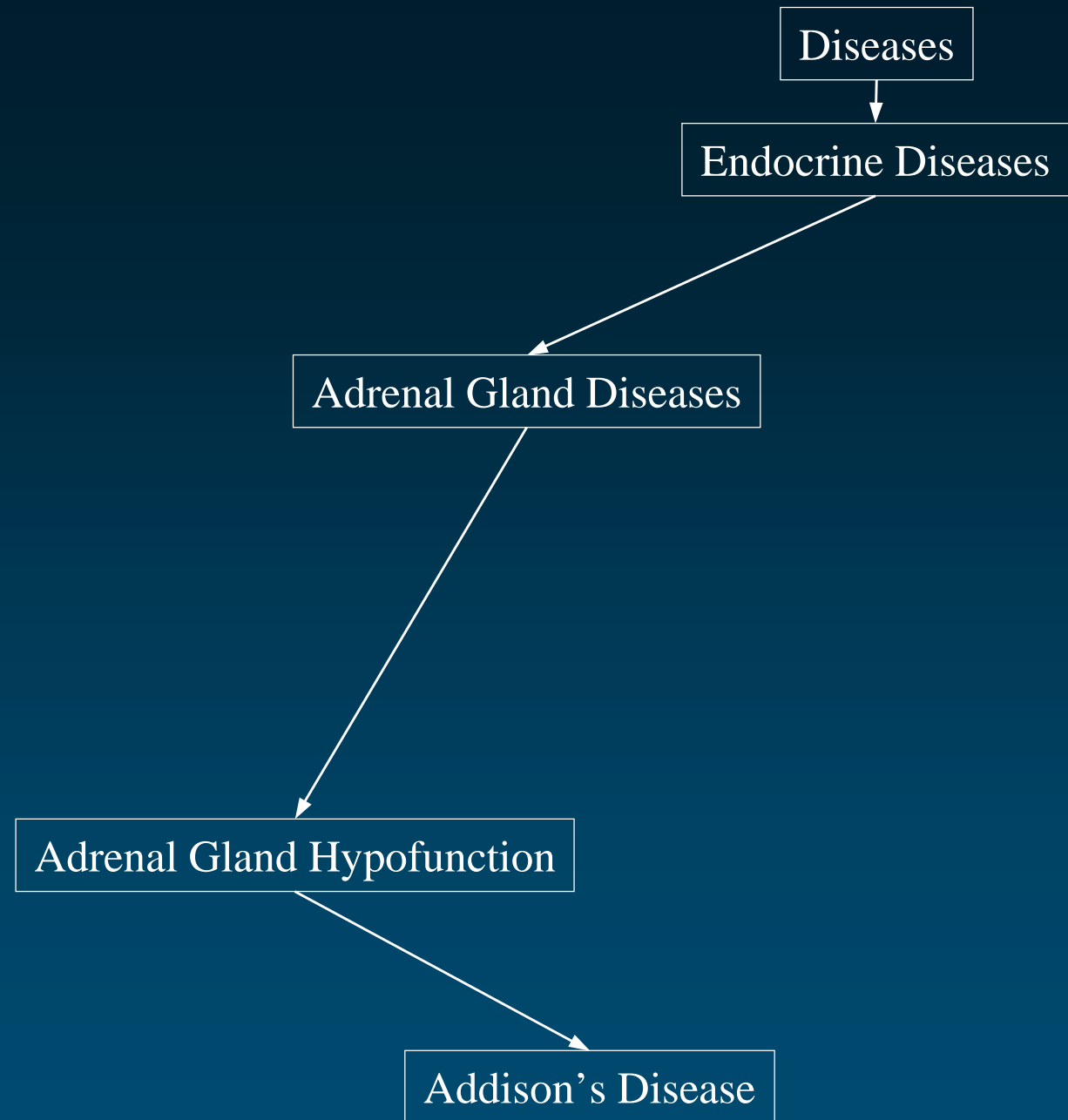
Distribution of depth max for 10,000 randomly selected UMLS concepts (2001) 5

Example Addison's disease

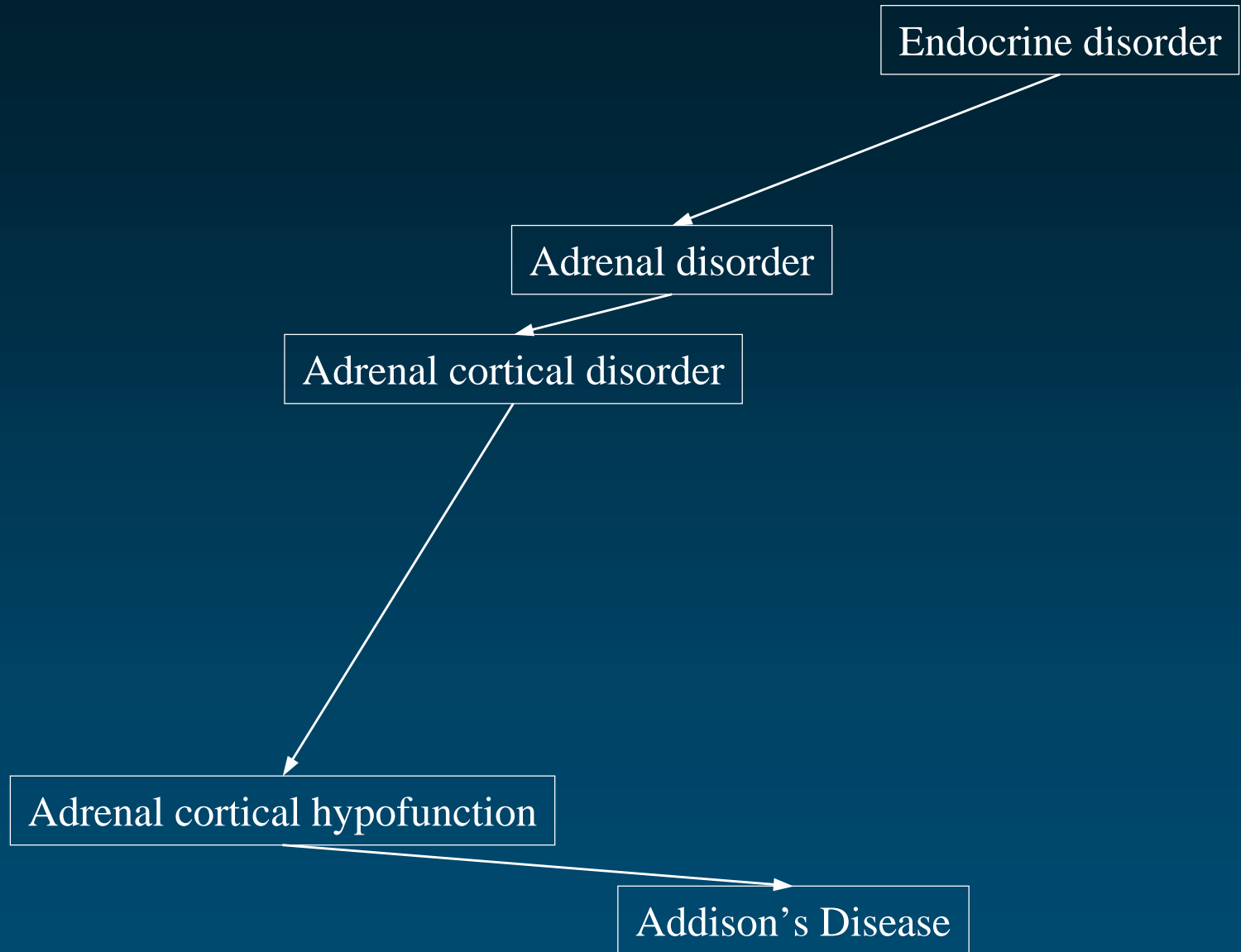
- ◆ Representation in several medical vocabularies
 - SNOMED International
 - Medical Subject Headings (MeSH)
 - Alcohol and Other Drugs Thesaurus
 - Read Codes (CTV3)
 - International Classification of Diseases
- ◆ Combined representation in the UMLS Metathesaurus

SNOMED International

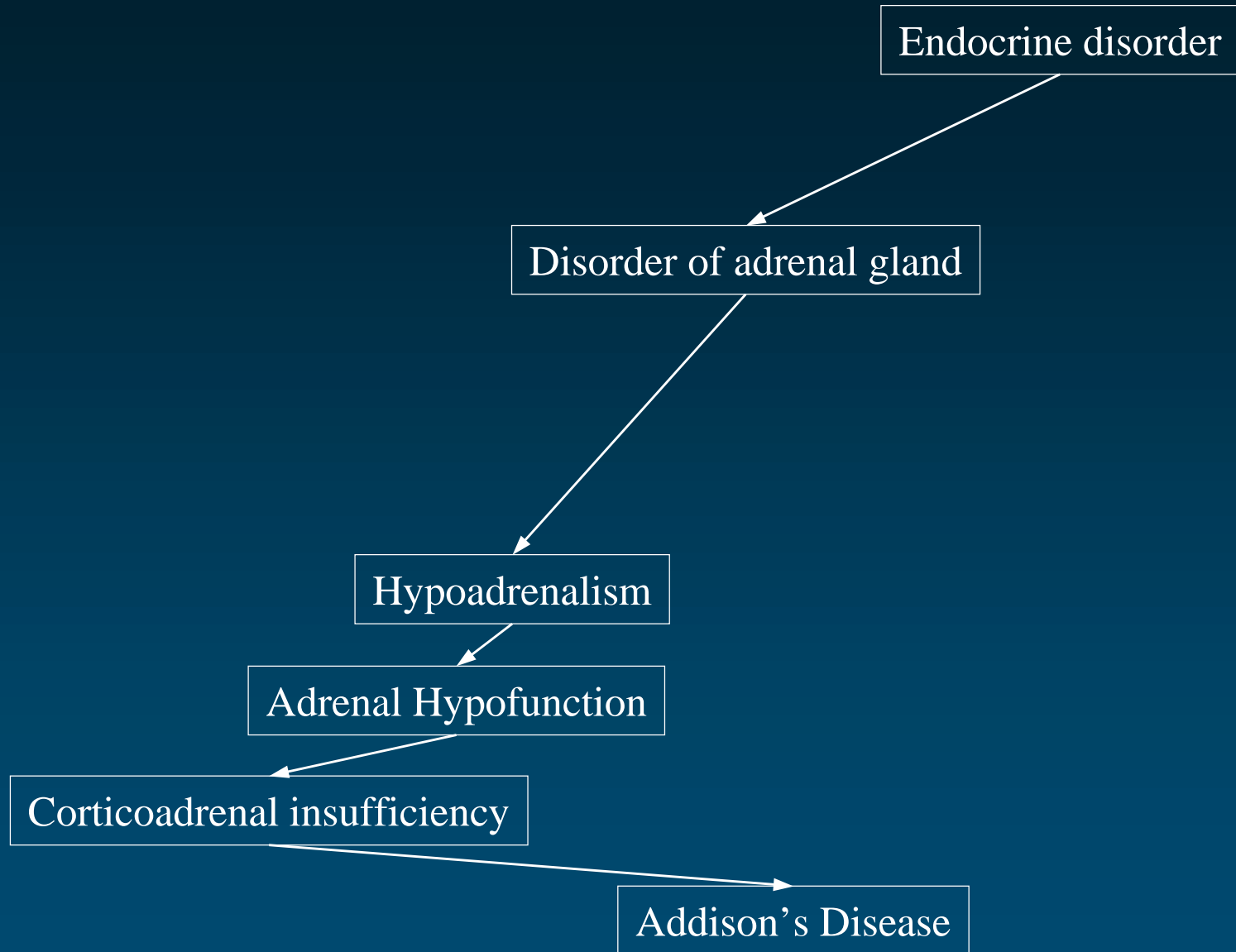




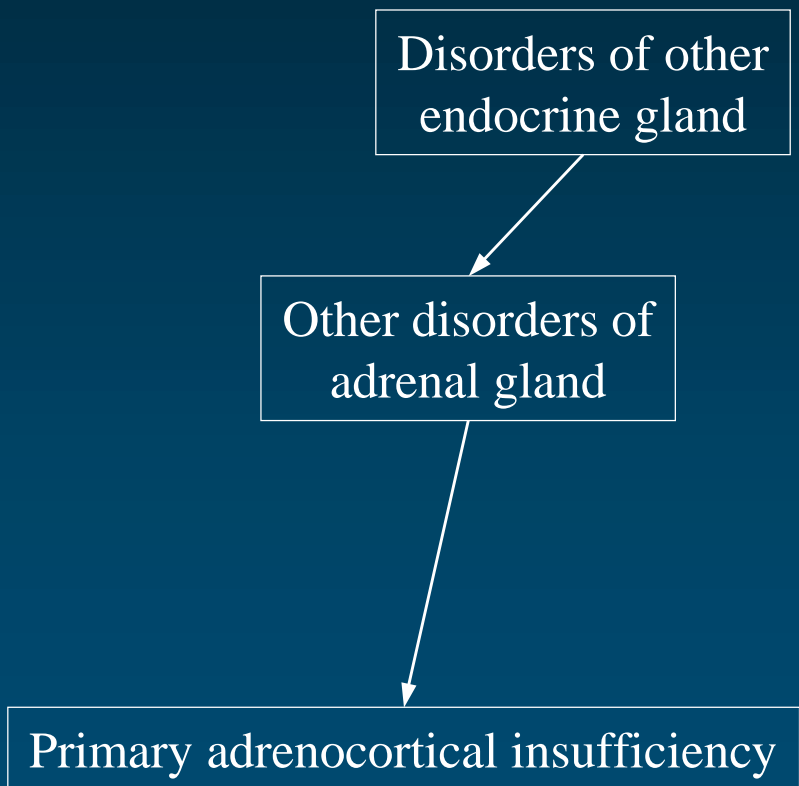
AOD



Read Codes



ICD-10



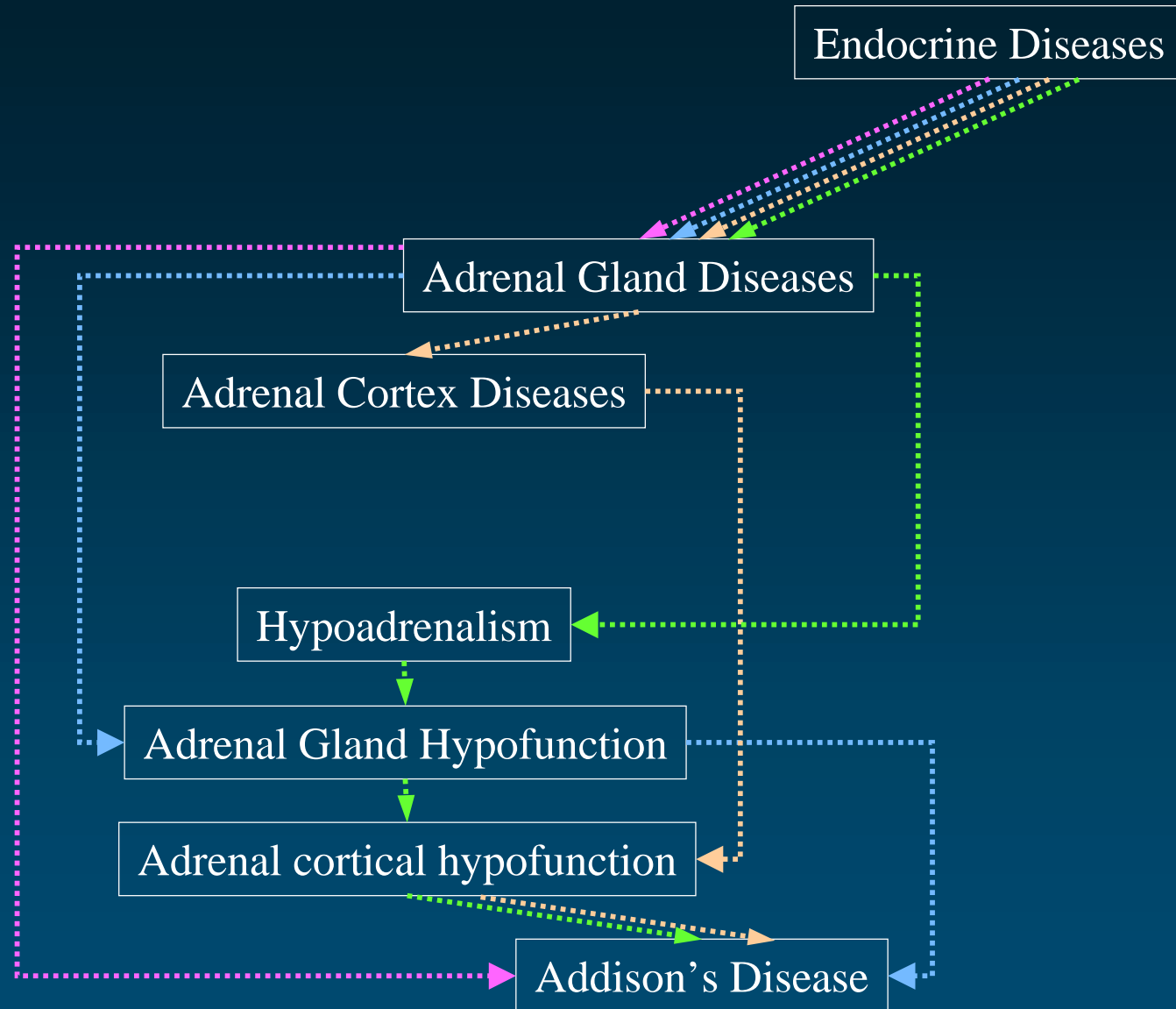
organize concepts

SNOMED

MeSH

AOD

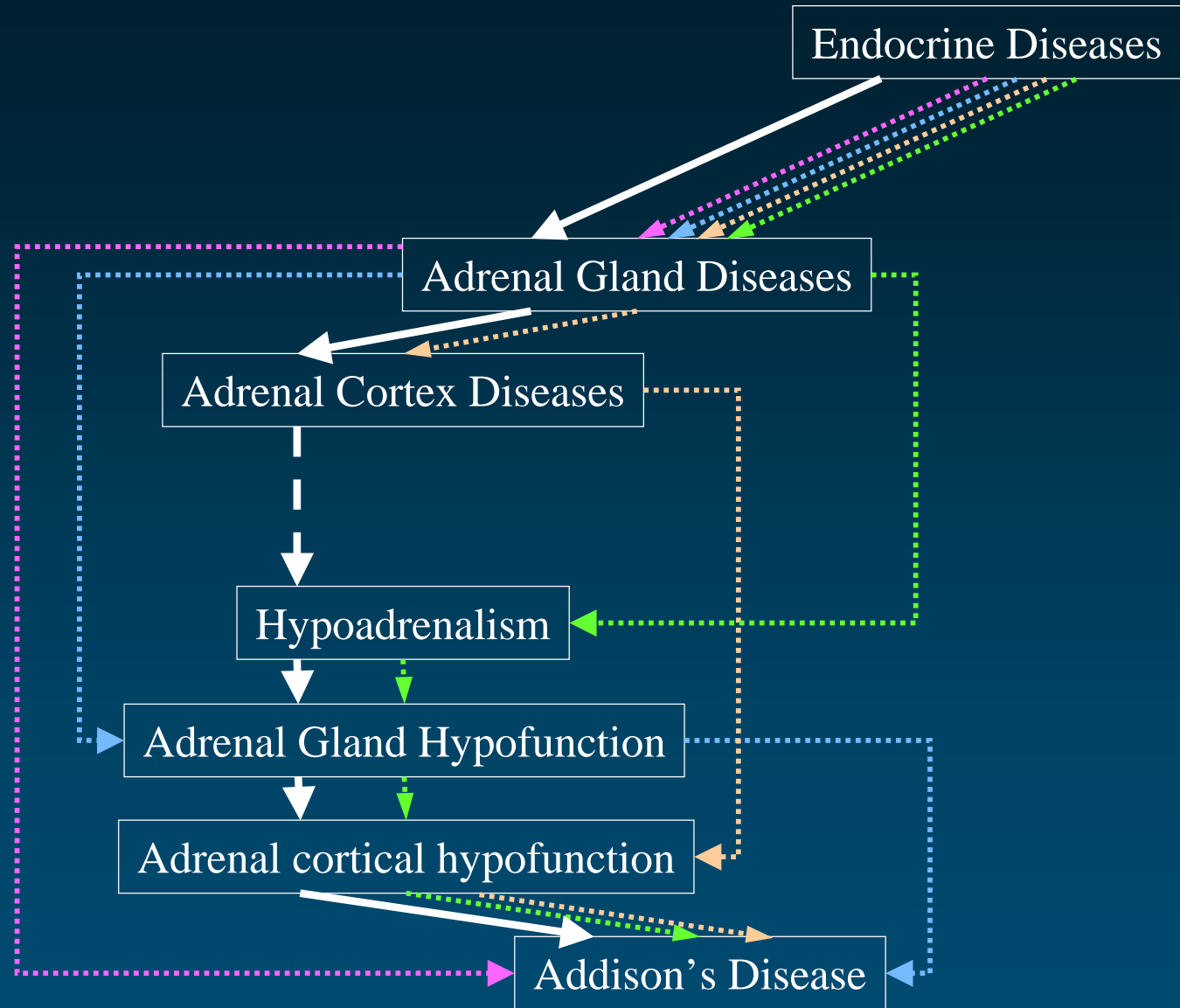
Read Codes

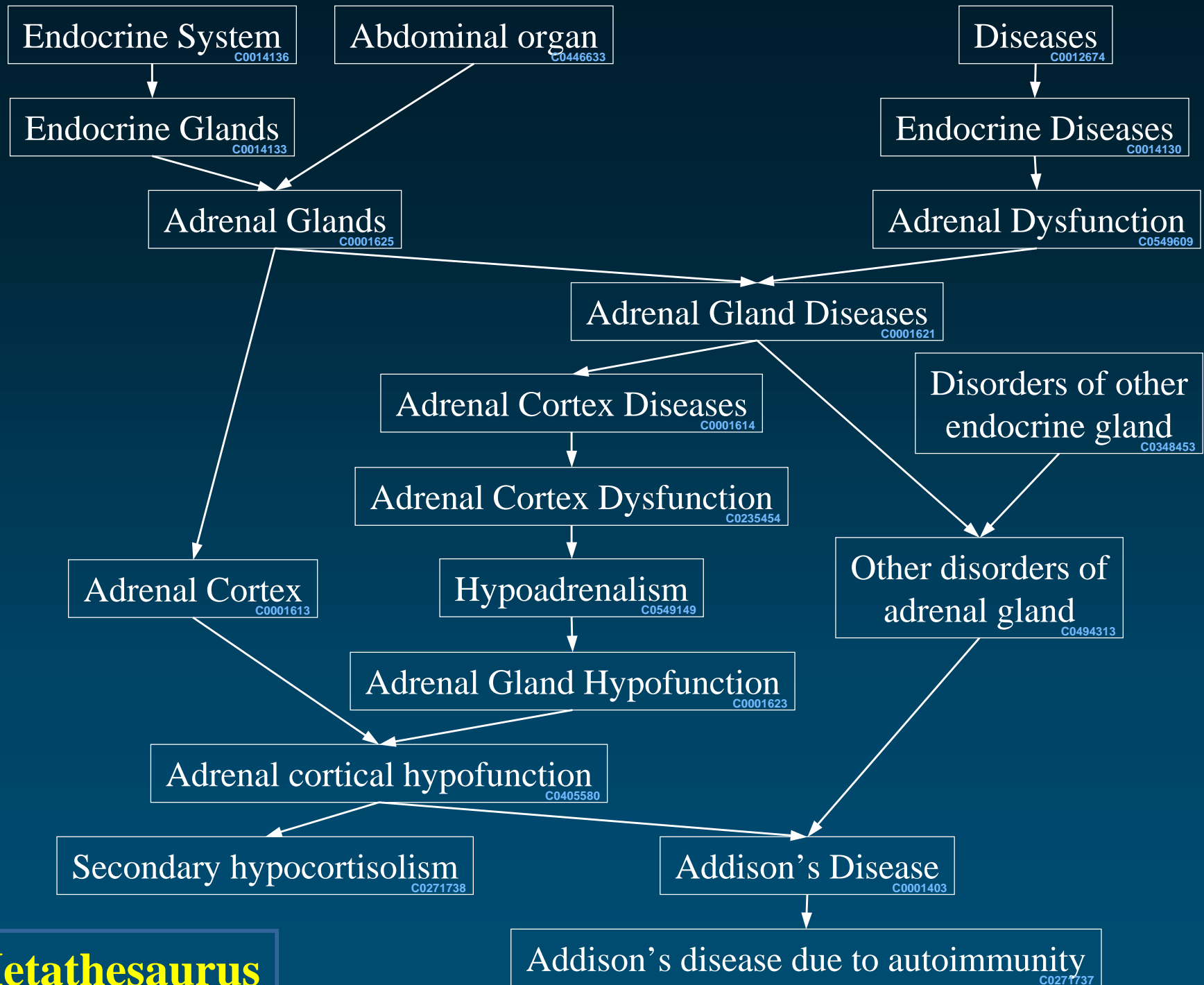


organize concepts

SNOMED
MeSH
AOD
Read Codes

UMLS





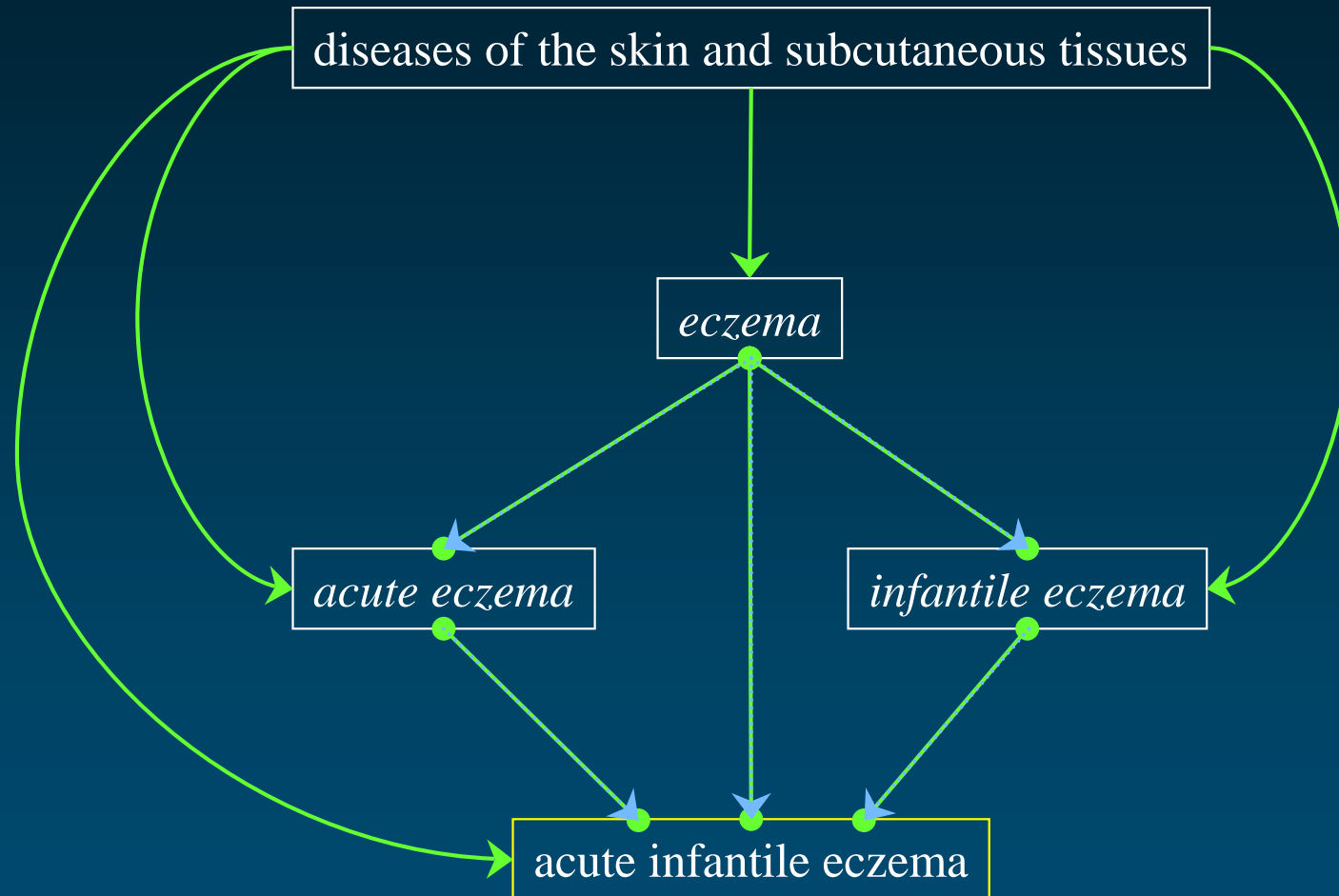
Metathesaurus

Granularity and synonymy

- ◆ Granularity may be limited in order to represent only significant differences among concepts (linguistic synonymy vs. clinical synonymy)
- ◆ When granularity is limited by design, it may not be possible to represent some hierarchical relationships

Example: Acute infantile eczema in SNOMED

Lack of structure within a source



Granularity and redundancy

◆ “Core” concepts

- Present in more than one vocabulary
- Essentially eliminates leaf nodes (structural equivalent)
- Also reduces the density of the graph
- Reduces the number of concepts dramatically ($> 80\%$)

Types vs. Instances

Types and instances Examples

◆ Types

- Liver *is a kind of* Organ
- Cirrhotic liver *is a kind of* Liver

◆ Instances

- Leipzig *is an instance of* City
- Barry Smith *is an instance of* Philosopher
- My liver *is an instance of* Liver
- This aspirin tablet *is an instance of* Clinical drug

Nothing can be *a kind of*

- Leipzig
- Barry Smith
- My liver
- This aspirin tablet

Types and instances Biomedical domain

◆ Types

- Terminologies
- Ontologies

◆ Instances

- Medical records
- Patient databases



used to
abstract away from
or reason about

◆ Classes

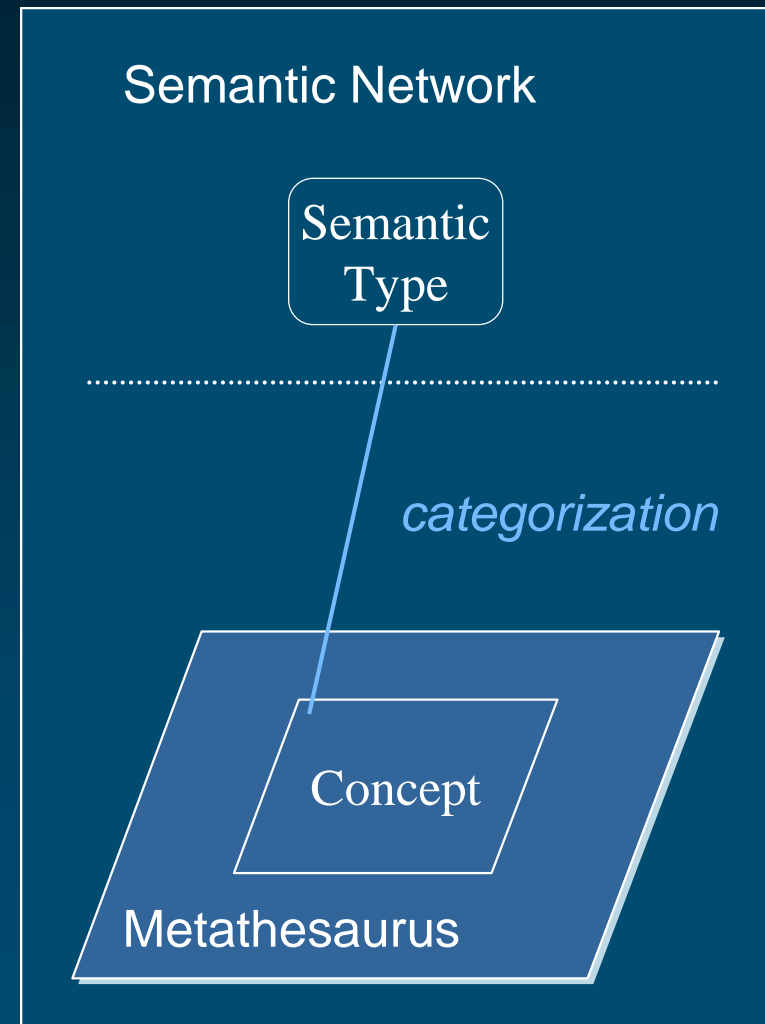
- Taxonomy
- Classification

Class:	Schizomycetes
Order:	Eubacteriales
Family:	Bacillaceae
Genus:	Clostridium
Species:	Clostridium botulimun

Types and instances UMLS

◆ Two-level structure

- Semantic Network
 - 134 Semantic Types (STs)
 - Relationships among STs
- Metathesaurus
 - 800,000 concepts
 - Inter-concept relationships
- Link = categorization
 - Often isa
 - Rarely is an instance of



Semantic Types

Anatomical
Structure

Fully Formed
Anatomical
Structure

Embryonic
Structure

Disease or
Syndrome

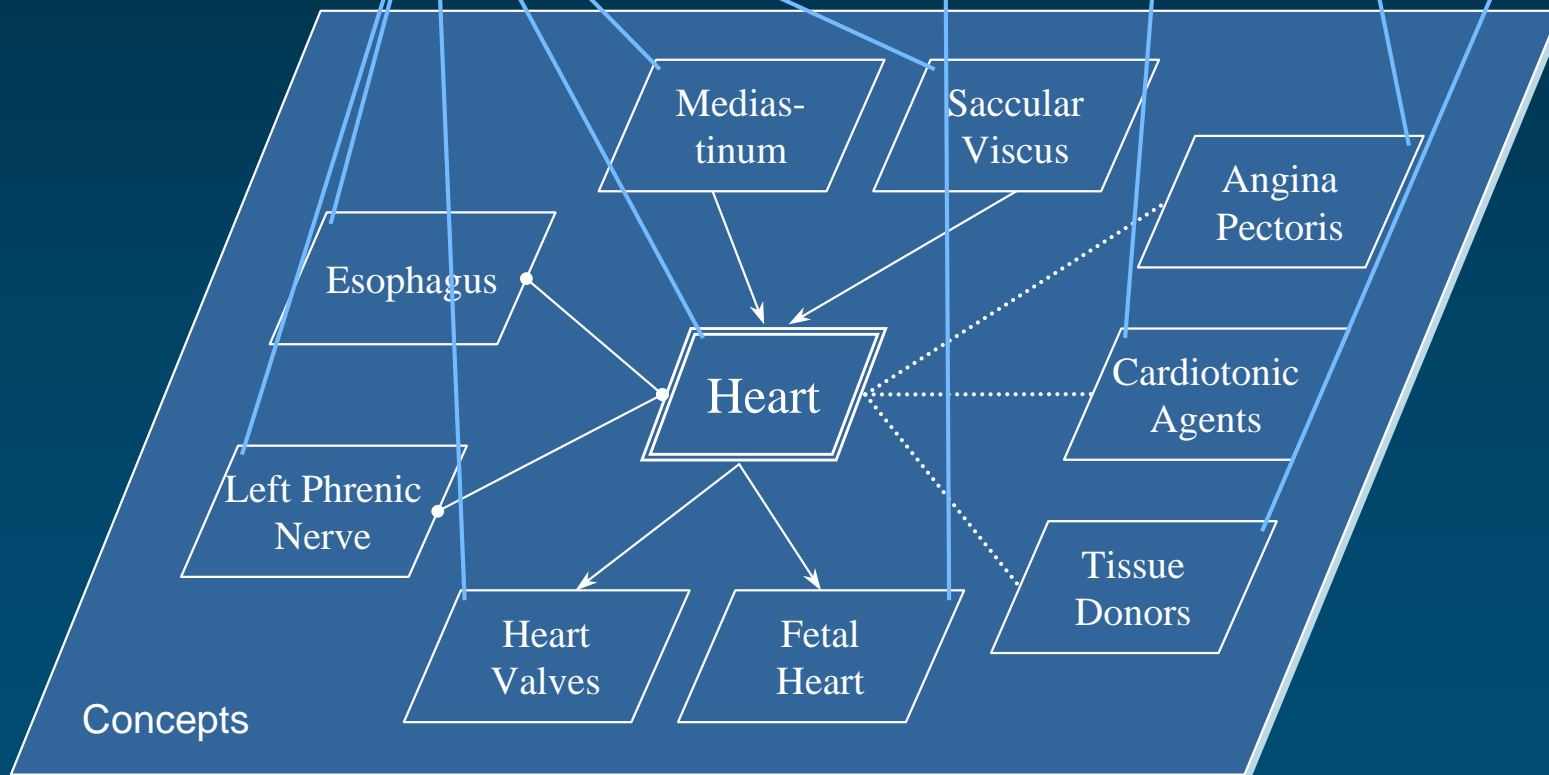
Body Part, Organ or
Organ Component

Pharmacologic
Substance

Population
Group

*Semantic
Network*

Metathesaurus



Concepts

Types and instances UMLS

- ◆ Essentially all types
 - Semantic types
 - Concepts
- ◆ Exceptions
 - Named geographic areas
Germany, Europe
 - Named laws
National Health Planning and Resources Development Act of 1974
 - Named intellectual products
Finnish translation of the Medical Subject Headings
- ◆ No explicit distinction
between *is a kind of* and *is an instance of*

Types and instances Identification

◆ Features of instances

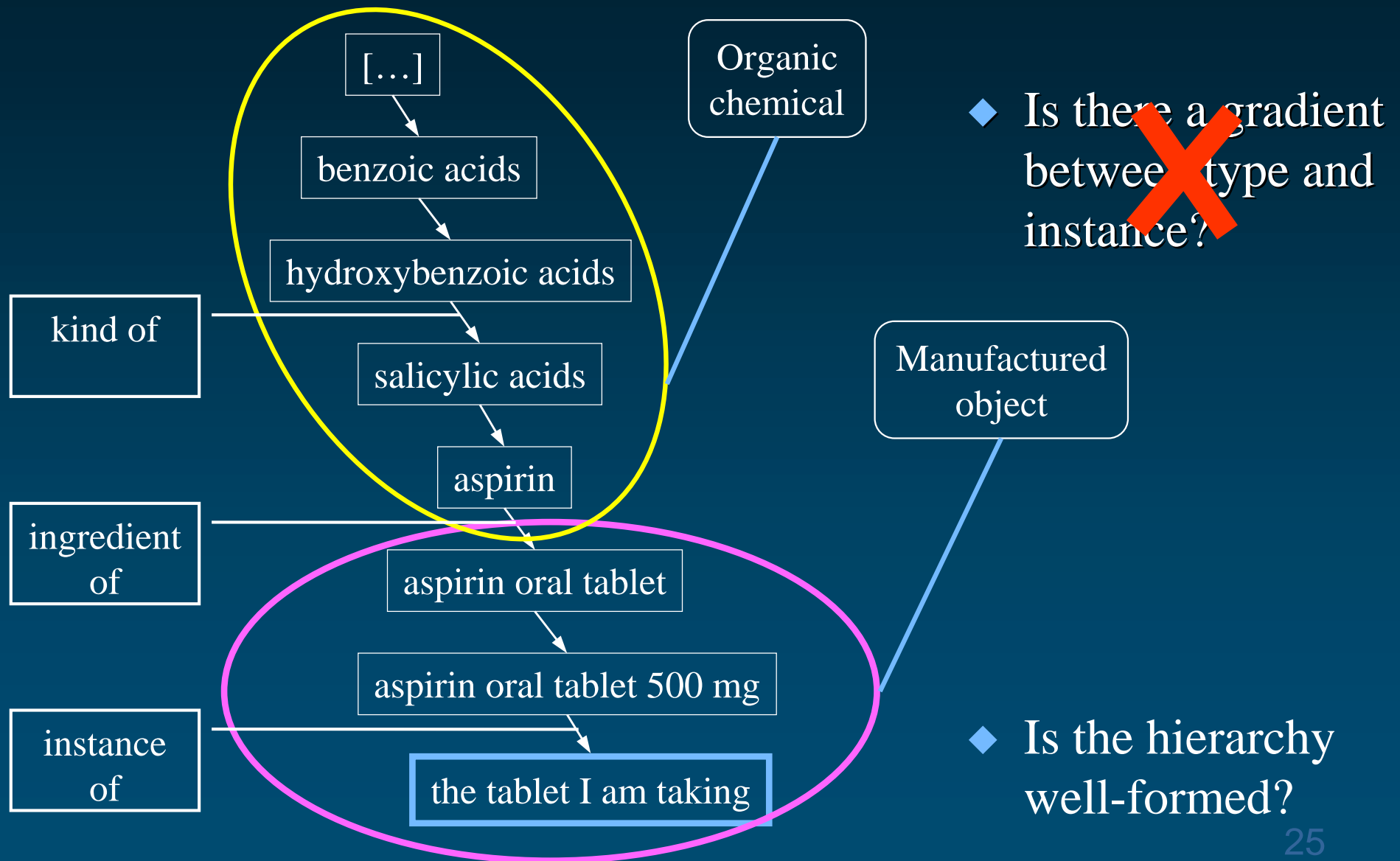
[E. Alfonseca, GWA 2002]

- Structural: often leaf nodes
- Morphologic: often capitalized
- Syntactic: usually not preceded by a determiner

◆ Applications

- Named entity recognition

Types, instances and granularity

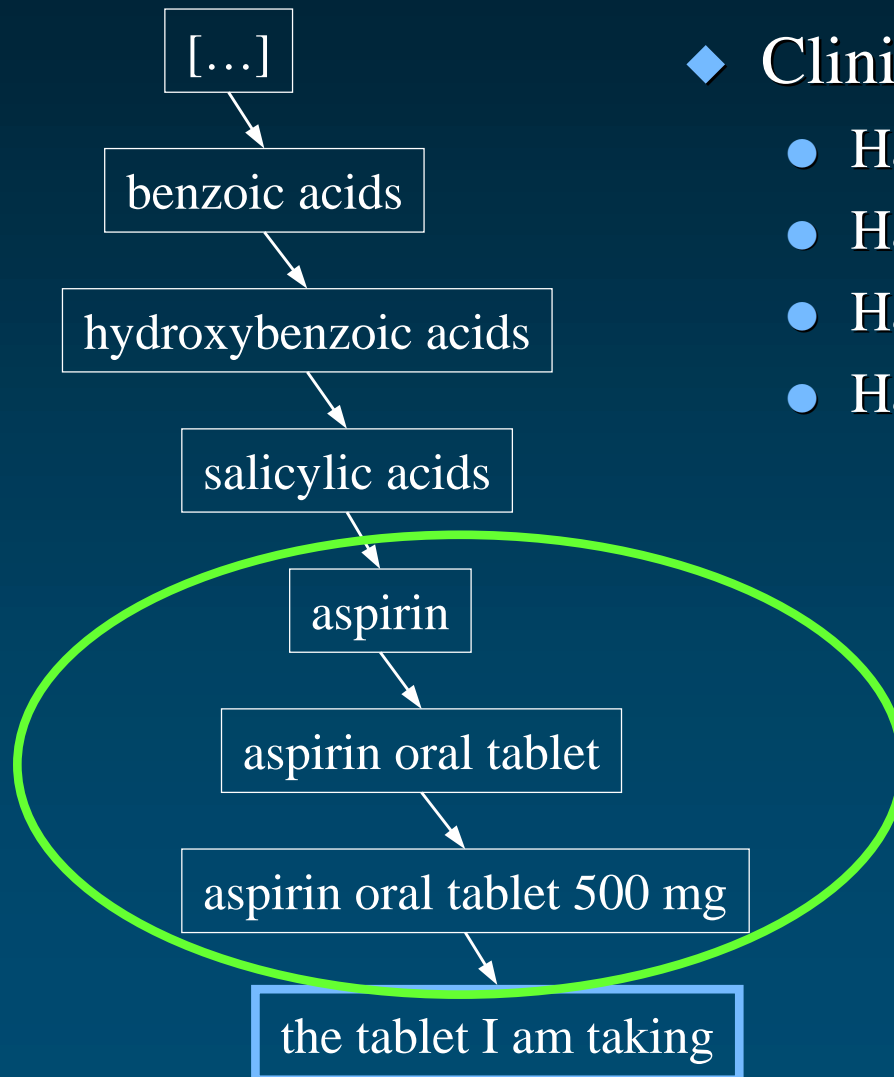


Types and instances Summary

- ◆ Biomedical ontology
 - Essentially types
 - Some classes (taxonomy)
 - Hardly any instances
- ◆ Instanciation: medical records
- ◆ Relationships: must distinguish between
 - Is an kind of (type-type)
 - Is an instance of (instance-class, type-class)

Ontology vs. Information model

Aspirin revisited



◆ Clinical drug

- Has ingredient(s) aspirin
- Has dosage 500 mg
- Has route oral
- Has form tablet

[RTM Drug Model, VA]

Other examples Medical procedures

◆ Medical procedure

- Action
- Anatomic site
- Instruments
- Approach

◆ Appendicectomy

- Remove
- Appendix
- Surgical instruments
- Open surgery

[MAOUSSC, GALEN-IN-USE, SNOMED-RT]

Other examples Lab/clinical results

◆ Lab test

- Component
- Property
- Time aspect
- System/Sample
- Scale
- Method

◆ Sodium measurement

- Sodium
- Serum concentration
- Point in time
- Serum/Plasma
- Quantitative
- N/A

LOINC “terms”: SODIUM:SCNC:PT:SER/PLAS:QN

[LOINC, Regenstrief Institute]

Ontology vs. Information model

◆ Ontology

- What: Meaning
- How: hierarchies, frames, description logics
- How big: often very large (hundreds of thousands of concepts)
- Access: through browsers

◆ Information model

- What: Structure
- How: UML diagrams
- How big: the model is often limited in size
- Access: “readable” diagram
- Populated with concepts from the ontology

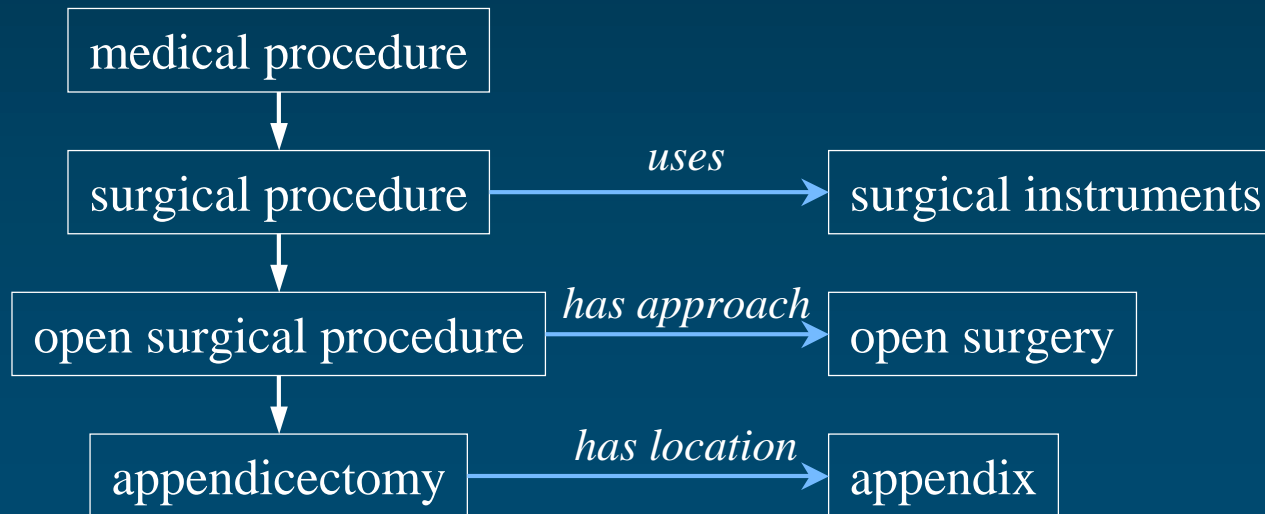
[A. Rector, MEDINFO 2001]

Although O. and I.M. may be equivalent

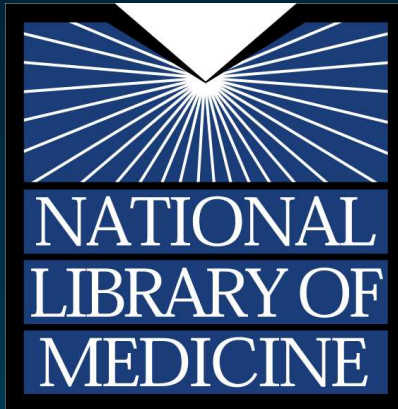
◆ Appendicectomy

- Remove
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(simplified
representation)



Contact information



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